

**Table 2. SETX Mutations Identified in Individuals with Ataxia with Oculomotor Apraxia Type 2**

<b>Exon</b>	<b>Mutation</b>	<b>Nt Position</b>	<b>Nt Change</b>	<b>Predicted Effect on Senataxin</b>	<b>References</b>
2	E65K	193	GAA>AAA	Missense; aberrant processing	Duquette et al 2005
5	M274I	822	ATG>AT?	Missense; aberrant processing	Asaka et al 2006
6	Frameshift after C292	879	DelT	Frameshift; truncation	Moreira et al 2004
	W305C	915	TGG>TGT	Missense	Moreira et al 2004
	R332W	994	CGG>TGG	Missense	Moreira et al 2004
8	P413L	1238	CCT>CTT	Missense	Moreira et al 2004
	H435R	1304	CAT>CGT	Missense; aberrant processing	Fogel & Perlman 2006
	H469R	1406	AAA>AAG	Missense; aberrant processing	Criscuolo et al 2006
	R788X	2332	CGA>TGA	Nonsense; truncation	Moreira et al 2004
	Q868X	2602	CAA>TAA	Nonsense; truncation	Moreira et al 2004
	Frameshift after L874	2622-2625	DelAGTT	Frameshift; truncation	Moreira et al 2004
	Frameshift after Q988	2966-2970	DelGGAAA	Frameshift; truncation	Moreira et al 2004
	Frameshift 1157X	3466	DelG	Frameshift; truncation	Criscuolo et al 2006
	R1294C	3880	CGT>TGT	Missense; aberrant processing	Asaka et al 2006
	R1363X	4087	CGA>TGA	Nonsense; truncation	Moreira et al 2004, Duquette et al 2005
	Q1441X	4321	CAG>TAG	Nonsense; truncation	Moreira et al 2004, Duquette et al 2005
	Frameshift after I1690	5070	InsT	Frameshift; truncation	Moreira et al 2004, Duquette et al 2005
	Frameshift after L1750	5249	InsT	Frameshift; truncation	Moreira et al 2004
	Frameshift after N1754	5264	DelC	Frameshift; truncation	Moreira et al 2004, Fogel & Perlman 2006
	F1756S	5267	TTT>TCT	Missense	Moreira et al 2004
12	A1945P	5833	GCA>CCA	Missense; aberrant processing	Fogel & Perlman 2006
12	L1976R	5927	CTC>CGC	Missense; aberrant processing	Duquette et al 2005, Fogel & Perlman 2006

Exon	Mutation	Nt Position	Nt Change	Predicted Effect on Senataxin	References
2	E65K	193	GAA>AAA	Missense; aberrant processing	Duquette et al 2005
12	L1977F	5929	CTC>TTC	Missense; aberrant processing	Fogel & Perlman 2006
	Eight exons deleted (16-23)	6106+3393_7101-22Del20648bp/ins25bp		Truncation	Criscuolo et al 2006
18	H2197R	6590	CAT>CGT	Missense; aberrant processing	Moreira et al <i>in preparation</i> , Fogel & Perlman 2006
18	P2213L	6638	CCG>CTG	Missense	Moreira et al 2004
22	P2368R	7103	CCA>CGA	Missense; aberrant processing	Chen et al 2006
22	Donor site	7199+5G>A		Skipping of exon 23 Cropped of 10% of SETX C-terminus sequence	Criscuolo et al 2006